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TRUST IT OR TRASH IT?
CREATING & ASSESSING GENETIC HEALTH INFORMATION

Meeting Date: September 22, 2009

PUBLISHED BY GENETIC ALLIANCE

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INTRODUCTION

Patients, parents, healthcare providers, and the public are inundated by a constant flow of information from myriad sources, both reliable and unreliable. This bounty of information is ever-expanding, along with opportunities to disseminate it online further and more quickly. Some of the information is credible, and some of it is bunk. We need a way to filter out the meaningless or harmful information so that we may heed the high-quality health information that can have a positive impact on our lives.

On September 22, 2009, the Access to Credible Genetics Resources Network, a cooperative agreement funded by the Centers for Disease Control and Prevention (Grant #5U10DD525036), held a meeting to examine these issues as they pertain to the creation and assessment of health information. ATCG and this meeting help individuals, families, and healthcare providers determine when to "Trust It or Trash It?"

HISTORY OF THE DEVELOPMENT OF THE TRUST IT OR TRASH IT TOOL?

Access to Credible Genetics Resources Network (ATCG) was a cooperative agreement focused on Duchene/Becker muscular dystrophy and Fragile X syndrome. Genetic Alliance—a network of thousands of organizations, including more than 1,000 disease-specific organizations as well as academic institutions, professional societies, government agencies, industry groups, and more—directed the grant activities in partnership with the following organizations:

National Center of Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention
Genetic Alliance (lead)
FRAXA Research Foundation
GeneTests
National Coalition for Health Professional Education in Genetics
National Council of La Raza
National Fragile X Foundation
Parent Project Muscular Dystrophy
University of Maryland School of Medicine

Goals of this project included:

- Defining evidence-based information,
- Creating a tool that both guides the creation of accurate, high-quality information and critically assesses the quality of information, and
- Creating an overarching system that allows this process to be replicated.

Objectives included:

- Determining a process to define evidence-based information, including some accessible metrics,
- Determining the background information, people, networks, and the process necessary for disseminating information once it has been defined as evidence-based,
- Creating a tool to analyze patient and provider information in a systematic way,
- Creating a tool to guide the process of creating complete, high-quality, accessible information, and
- Identifying the components needed to create a robust, coordinated system for repeating this process with other health conditions, beyond Duchene/Becker muscular dystrophy and Fragile X syndrome, and even beyond rare conditions.

Audience for the Project

The ATCG audience is anyone who interacts with health information: diagnosed individuals, their families, and their care providers, including physicians, nurses, physician assistants, physical therapists, occupational therapists, educators, directors of health plans, etc. Though the project focuses on health information surrounding rare genetic conditions, it is also applicable to more common health information. We realize that the vast majority of healthcare providers are not experts in genetics, so disease-specific and genetic information developed for consumers is often useful for providers, as well. By defining "evidence-based information," and by characterizing information based on this definition, we hope to provide an accessible way for our entire audience to determine which information should be trusted.

Principles of ATCG

The ATCG process, including the culminating meeting, progressed according to the following principles:

1. Work is transparent and immediately available for others to use or comment on,
2. The evaluation process is transparent enough that others can understand the process and make their own judgments as needed,
3. The field of evidence-based medicine informs this project,
4. Models from other fields are examined and integrated where possible,
5. Other projects and programs in information, genetics, resources, single gene disorders, common conditions, and genetic testing and screening are examined and their leaders are consulted and invited to participate,
6. The information assessment systems created are patient-focused and replicable, and
7. Our results are meant to be practical – they will not require that "further research be conducted," nor will we wait to disseminate even preliminary findings, plans and results; all information will be distributed multiple times throughout the project.

Evidence-Based Information

The field of evidence-based "information" is relatively nascent, and cannot solely rely on the concepts developed by the field of evidence-based "medicine." For information to be credible, the process of developing information must be completely transparent. Credible sources, which are those based in science and medicine, are the foundation for high-quality information. However, we determined that the utility of even high-quality information is relative to the individuals using it. Most primary care providers have limited knowledge of genetics so information on genetics written at a high level might have low utility for them[1]. Furthermore, there is not enough accurate information about single gene disorders readily available for providers or patients.

At the start of our project, we considered the Agency for Healthcare Research and Quality (AHRQ) scoring system for evidence-based medicine as a potential set of metrics to determine what is evidence-based information[2]:

Agency for Healthcare Research and Quality
Scoring System for Evidence-based Medicine

A: randomized controlled clinical trials,
B: nonrandomized case-control studies,
C: nonrandomized cohort study,
D: clinical report with nonrandomized historical comparison groups,
E: case report or series,
F: expert consensus,
G: subject review subsuming multiple categories A-Et

We concluded that information for most single gene disorders will not be able to meet the requirements of A or B, and often cannot even meet the requirements of C-F. It appears that the AHRQ system is not easily applicable to single gene conditions because of the rarity of these diseases, and also because it is focused on medicine, not information.

Evidence-based medicine is largely based on quantitative information that is critical to the definition of evidence. When considering evidence-based information, as opposed to medicine, *qualitative* information must also be integrated, for example, details about a patient. Negative results and "no findings" results are generally not published, but are important when considering and producing accurate information. Another concern is that single gene disorder information may be accurate but incomplete, which makes it less than completely reliable. As with evidence-based medicine, information should be continuously examined and reevaluated as the field advances. In the course of the project, we determined that the term "evidence-based information" did not wholly capture the essence of what we were seeking. Instead, we found that terms such as "accurate", "reliable", and "high-quality" better described the types of information we sought to capture.

Individuals must have accurate and complete information to make informed health decisions, yet consumers and providers face many challenges in accessing credible information The wealth of available information on both rare and common genetic disorders is constantly expanding; some information in both print and Web-based resources is plainly inaccurate[3,4,5]; many healthcare providers have limited knowledge of and experience with genetic disorders[6]; and the public has a generally low health literacy rate[7].

Which Stakeholders Should Be Involved in the Creation and Maintenance of Information?

It is important to build standard methodologies for information development that can be applied to all conditions. The *Trust It or Trash It?* tool—a guide to creating and assessing educational information for both rare and common health conditions—helps establish a level of consistency and credibility for consumers. Throughout its development, we integrated a great deal of feedback from many sources to improve the tool's sensitivity and utility. ATCG partners conducted a review of the current literature, available tools, and guides; invited iterative discussion with and review by multiple stakeholders; and held focus groups with advocacy group members, members of the general public, medical students, pediatric residents, and curriculum developers. Additionally, the ATCG National Advisory Committee and condition-specific scientific advisory groups reviewed the tool.

Over the course of the project, ATCG partners and advisors determined that the following areas of information constitute comprehensive coverage of a condition: basic information on the condition, including features, etiology, populations affected, and research; medical care, including choosing health professionals, diagnosis, symptoms, treatments, and financial considerations; developmental issues, including changes over time, behavioral issues, and expectations; and family issues, including impact, changes over time, and risk communication. All these areas must be regularly reassessed because information changes rapidly, becoming outdated and often incorrect.

Disease advocacy organizations (DAOs) are in the ideal position to keep track of this constantly changing suite of information. Because they are dedicated to specific conditions, DAOs are alert to advances and aware of the state of the science and can therefore verify what is the most current information about their particular conditions. This attentiveness serves to make DAOs excellent stewards of condition-specific information. In addition, DAOs encourage multidisciplinary participation in their disease-specific work, so many experts and stakeholders can be involved in information development: disease-specific advocates, patients and family members, healthcare and allied health providers, researchers, industry representatives, health educators, and others. This multispecialty/multidisciplinary approach is critical to ensure information is high-quality, complete, and applicable to multiple audiences.

This project did not grant imprimaturs to creators of information on genetic disorders. Instead it produced the *Trust It or Trash It?* tool that allows the user to judge for him or herself the quality of a resource. This approach fosters critical thinking by individuals and is sustainable for the tool and information developers; it would be unfeasible to attempt to stamp a seal of approval on all high-quality resources.

MEETING WELCOME AND CONTEXT

Sharon F. Terry, MA, Genetic Alliance, welcomed participants and pointed out that the range of organizations collaborating on the ATCG project represents the people for whom this information is critical: disease advocacy and community-based health organizations as well as health professionals. Terry conveyed what needs to occur so that both patients and providers have access to high-quality health information.

There is now an overabundance of information for healthcare providers and individuals and families. Accelerated research produces a constant stream of information, and new media by which to communicate this information electronically regularly arise. It is our task to offer the public a tool by which they may determine whether to trust the information they encounter.

THE SECOND “5,000 DAYS OF THE INTERNET”

In 2007, Kevin Kelly created a video clip to summarize the evolution of the Internet and predict changes yet to come[8]. ATCG's work to assess information has exciting possible implications. Connections are now made between Web pages by sharing url's through links. A link on one page brings you to another page, but not necessarily to the specific piece of content on that page in which you are interested. There may be future opportunities to link ideas to each other within Web pages, connecting the Internet on the level of actual ideas, or topics, rather than whole pages. These new connections generated by Web 3.0, the next generation, customized Web experience, will offer more personalized health information. The Web will continue to evolve, using a person's search history to anticipate and deliver new information that reflects their interests.

KEYNOTE ADDRESS: LINKING TECHNOLOGICAL POWER TO THE ASSOCIATIVE POWER OF THE HUMAN MIND

Recently diagnosed individuals and their families often turn to the Internet for answers, but how do they make sense of all the information and decide what is accurate? How can they get the maximum payoff from the Internet, a powerful and evolving tool? Nickolas Barris, Concept Web Alliance, described new ways to harness the Internet to maximize information delivery. Tapping into the power of the human mind and technology will mean not only an increase in high-quality information but a decrease in redundant information as connections generated on Web 3.0 evolve.

Barris has expertise in Web 3.0 concepts and has a personal interest in developing resources on genetic conditions. His wife's family is affected by a rare genetic condition that occurs in fewer than 200 people worldwide: Hallermann-Streiff syndrome. Recently, just months after running a marathon, his father was diagnosed with a rare blood cancer and has deteriorated rapidly. “One thing I've learned about dealing with my father's terminal illness is that we are sprinting a marathon.” To a patient or a caregiver, timely, near-instant access to the most cutting edge data and educational information on a genetic condition is critical.

Today, entering “stroke” into a search bar yields more than 60 million hits. A patient seeking information may quickly become overwhelmed by the magnitude of results, and frustrated seeing redundant and inaccurate pages. In the future, information on the Web will be better organized to minimize redundancies while increasing the rate of information delivery. The mass of information about a condition can be “curated,” edited into discrete concepts tagged with unique identification numbers to link similar concepts to each other. This identification will drastically simplify the user experience: information will be organized into a taxonomy that preserves all available information but structures it to eliminate redundancy. As information is increasingly organized (eliminating the chaos of “search engine overload”) we will move toward a finite number of related concepts.

The chance for something unexpected and novel to emerge still remains, even with this intensification of organization. Barris explained that his company, Concept Web Alliance, provides online connections to different concepts in different countries, which might not have been disseminated otherwise. These connections can create “a spark,” a sudden insight that two previously unconnected concepts are, in fact, related. This has profound implications for advancing knowledge about rare genetic conditions, for bringing esoteric information into the mainstream and for discovering meaningful links.

The success of Wikipedia shows how human minds and technology can work symbiotically to advance knowledge more quickly than could be accomplished through person-to-person collaboration. Extending the wiki data model to life sciences holds great promise for extending the reach of high-quality information. The goal is not only to eliminate redundancies but to disseminate information more quickly, allowing thinkers to add knowledge instantaneously.

While we are making tremendous strides in connecting technology and the human mind, a familiar problem remains: how to ensure the quality of information being disseminated. The ability to connect people with information is meaningless if the information itself is not credible and accurate. This has been a problem for Wikipedia and, indeed, for all wiki data models.

Participants were invited to ask Barris questions and make comments. A summary of most of these comments, which focused on the user experience with the new technology and the dangers it poses, is below.

- Patients are particularly vulnerable Web visitors, because they often seek new information about a diagnosis, symptom, treatment, or another aspect of living with a disorder in the hopes of improving their situation. Patients often endeavor to learn as much as possible about a genetic condition, and therefore should be recognized by experts as a unique audience.
- Children should be taught how to use new technologies as they emerge, and be aware of the wealth of misleading, potentially harmful information available.
- Even experts with strong credentials and backgrounds sometimes disagree with each other and claim that their answer is right. These discrepancies should be communicated to users in a way that minimizes confusion.

PANEL DISCUSSION: QUALITY

Miriam Blitzer PhD, University of Maryland School of Medicine, introduced the panel on how to assess the quality of information, moderated by Andrew Faucett, MS, CGC, Emory University. Panelists included 1) Jeffrey Botkin, MD, MPH, University of Utah; 2) Roberta Pagon, MD, GeneTests; 3) Nickolas Barris, Concept Web Alliance. Blitzer explained that consumers and healthcare professionals need the right tools to examine the quality of information and make informed decisions.

How is Quality Assessed in Genetics and Health?

Jeffrey Botkin raised ethical questions about the genetic testing field and offered strategies for using information derived from tests to achieve desired outcomes.

Botkin set the context in which new information about genetic conditions is emerging, referring to “a flood of information” created by a confluence of three sources:

- The Human Genome Project
- Enhanced interest of clinicians
- Better information sharing, thanks to new technologies such as those described by Barris

Given this “flood of information,” we should allow ethical considerations to guide the way we gather information. Researchers should carefully define the range of outcomes of genetic tests before implementation. Botkin gave two examples of tests where desired outcomes must be weighed against the technical ability to perform the test: prenatal diagnosis and newborn screening (NBS). For each test, what is the desired outcome? For whose benefit is the test performed? Traditionally such tests were thought to benefit the individual screened, but today these tests are additionally being used to benefit parents, by sparing them a “diagnostic odyssey,” and the healthcare system, which allocates disproportionate resources to patients suffering from devastating genetic conditions.

Now that some genetic tests are available for online purchase, the ethical questions around genetic testing are even more pressing. Some online genetic testing services allow consumers to learn about their ancestry and health risks due to genetic predisposition. Botkin worries that direct-to-consumer genetic testing may lead to patients drawing inaccurate or incomplete conclusions about their health and risk for disease.

Furthermore, genetic tests themselves are becoming rapidly sophisticated, and ethical considerations about the use of the new information are not being addressed at the same rate. To highlight the importance of ethical considerations in genetic testing, Botkin points to the acronym ACCE for the quality of information gathered from such testing: A = analytic validity, C = clinical validity, C = clinical utility, and E = ethical, legal, and social aspects. The "E" concerns how well the test achieves desired outcomes.

Genetic testing has become increasingly sophisticated, yet three inherent problems remain.

- Assessing the impact of genetic test results is difficult because it is a "moving target" with regard to the type of information that patients or parents have over time. The classification of conditions may change, the patient may respond in an unexpected way, a new category of patients may emerge over time, etc.
- There is always lag time between receiving an individual's genetic test results and understanding that person's ultimate health outcomes, including how long the patient lived, how severe the symptoms are at the end of life, etc.
- The rarity of some genetic conditions makes it challenging to collect enough information from comparable subjects.

One positive counterpart to these problems is the concept of an unexpected positive result that can arise from genetic tests: what Botkin calls "boxcar information." This is information accidentally learned about a condition by testing for a different condition.

To summarize, Botkin described a two-part procedure to create high-quality genetic tests. First, work "backwards," that is, approach each test with the desired outcome in mind. Then, assess how well the test produces this outcome using the ACCE standards outlined above. However, implementing this two-part procedure across the field of genetic testing would require new research structures.

Practice: A Model of the Collection of High-Quality Information

Patients are not the only people who turn to the Web for instant up-to-date information about genetic conditions. Roberta Pagon described a Web-based resource designed to "help a healthcare provider get through a first encounter with a patient with a disorder for which they are not an expert." GeneTests provides "documents that are written to promote the integration of genetic testing into patient care." This website is "a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to interested persons." Once restricted to healthcare providers with special access, this Web-based resource is accessible without restricted access. It is, however, deliberately written with language more suited to doctors, its target audience, than consumers. GeneTests is also closely linked to a directory of labs offering specific genetic tests.

Healthcare providers should not have to filter through tangential information to get the information they need. The busy physician with little genetics background, treating many patients with a wide variety of conditions, needs access to the most cutting edge data on a condition, fast. he needs to know:

- What condition does my patient have?
- What is the prognosis of patients with this condition?
- Is anyone else in the family at risk?
- What are the next steps? This question includes treatment, preventive care, and counseling.

Pagon described quality control at GeneTests. Invited or volunteer experts submit GeneReview articles, and, as in academia, their work is then submitted for peer review. Authors of GeneReview articles must include one member of the target audience and a clinician. Articles must not only be accurate and up-to-date, they must meet the needs of the busy physician who is not a genetics expert. In this regard, editing should adhere to consistent stylistic conventions that allow healthcare providers to quickly navigate articles to find the appropriate information. Genetic tests referenced in the article must appear in the GeneTests laboratory listing.

Vision: Where Are We Going?

The keynote speaker, Barris, returned to talk more specifically about laying the foundation for Web 3.0. He described a collaboration with Wikipedia as well as an excellent website run by the Duchenne muscular dystrophy community, www.duchenneconnect.org.

Barris' collaboration with Wikipedia involves working to increase expertise, specifically enhancing their biomedical articles to include "expert views," thus making this well-established, consumer-friendly website more credible.

Work with the Duchenne muscular dystrophy community involves exploring consumer-friendly aspects of the Web that may be out-of-sync with the scientific perspective. Reconciling consumer needs with the scientific perspective will be an important quality of future Web-based resources for patients and families seeking health information.

- Patients want a place where they can read and share stories and opinions of other people going through the same thing, whether they are "accurate" or not.
- Blogging is a well-established way that patients and caregivers build a sense of community. However, blogs can be unpredictable and may contain misinformation, either intentionally or unintentionally.
- Cutting edge research should be included on a patient community website, but its editors should remember that new research may run counter to deeply ingrained beliefs of that community.
- Terms should be defined clearly and used consistently, both on community-building sites and those used by researchers.

Overall, Barris sees blogs and other consumer-friendly websites as invaluable tools for individuals and patient advocacy groups to become more informed and to assess the quality of health information.

Discussion

Moderated by Andrew Faucett

Conflicting Points of View

If Expert A says a particular treatment is effective and Expert B says it is ineffective, what can patients and healthcare providers take away?

First of all, it is imperative not to bury disagreement: the fact that there is disagreement between two qualified and experienced people is critical information, and both patients and providers are entitled to know this. When experts' views conflict, consumers should try to answer the following questions: Is there a financial conflict of interest underlying the divergent points of view? And, what are the qualifications of each expert—is one of them an "outlier," someone who consistently advocates views that are perceived as unorthodox? Is one of them a seasoned practitioner who has a new view that threatens the status quo? A conflicting point of view on a topic is a sign that consumers need to seek more information rather than choosing sides without further investigation.

Disseminating Information via Web 3.0: Speeding Up. Vs. Slowing Down

For patients faced with progressive conditions and for busy physicians trying to educate themselves about rare genetic conditions, the timeliness of access to current information is critical. On the other hand, it takes time to evaluate the data before any new medical treatment can be considered safe. This heightens the problem of quality control. There is more genetic information than ever, and Web 3.0 will allow the information to be disseminated more quickly than ever. However, the process must still be slow enough to allow for thorough evaluation of the accuracy of the information being disseminated. Web 3.0 is inherently built for speed; the Internet will identify connections and make links between concepts before humans have a chance to vet the connection. Pagon emphasized the danger in allowing computers to display medical information without human review: It "cannot be accepted." The audience understood that "rushed judgments can lead to wrong conclusions." Despite the threat of a patient or physician not learning something in time to help that patient, taking the time to allow for the appropriate review of new data by experts over time will ensure greater patient safety.

It is not only the inherent speed of Web 3.0 that creates pressure to release information prematurely. Pagon pointed out that most commercial labs use "aggressive marketing" techniques, but very few enlist medical experts. Furthermore, the real-life clinical utility of new treatments may not be a high priority for the labs that offer them. For a commercial enterprise, it can be better to release a new product fast than spend time to ascertain if it is safe and effective.

As Web 3.0 evolves, there needs to be a mechanism for human review of computer-generated connections between concepts, even at the cost of slowing down the dissemination of new information about genetic conditions. One member of the audience expressed a viewpoint: "We may be slow, but human health is not a commodity that we can afford to take risks on."

Patient Need and Scientific Data

Botkin stated, "there is no evidence for the treatment of genetic conditions." He meant that there is a disconnect between the raw data that scientists collect in labs, which leads to a greater understanding of the condition at a cellular level, and the medical needs of the patients. A breakthrough in the understanding of a condition at a cellular level does not directly yield effective treatment. As the ATCG project asserted, evidence-based medicine therefore does not apply in the case of rare genetic conditions. "We know very little in medicine. As you look at genetic testing literature, the level of high-quality information is really very low." Looking to the future, another panelist explained more optimistically: "We are at stage one of a ten-stage process."

Therefore, "credible genetics information (not medicine) is the focus" at this point in time. Nonetheless, the dearth of information about the treatment of genetic conditions "does not mean that people should be paralyzed." Physicians need to be explicit about exactly how much is known so that families understand the information available about the condition affecting their loved one.

Web 3.0 for Consumers and Professionals

The audience discussion turned to exploring how some barriers to information may need to be preserved, even as the next 5,000 days of the Internet brings unprecedented access for consumers of scientific information. GeneTests may be accessible to the public, for example, but there are still areas where professionals need to speak directly to other professionals without public scrutiny. This is not only to protect the public from data that has not yet undergone the time-consuming vetting process by experts, but to protect researchers investigating a hypothesis. To conduct their work, researchers need to experiment and conduct trials that may fail or produce promising results that ultimately cannot be validated. Public scrutiny of this scientific work may be harmful both to patients and to the research process investigating health conditions.

PANEL DISCUSSION: EXPERTS

Carol Greene, MD, University of Maryland School of Medicine, introduced the question "what is expertise?" The panel, moderated by Nancy Green, Columbia University Medical Center, included 1) Christine Vocke, MS, PXE International; 2) Howard Levy, MD, PhD, Johns Hopkins University; and 3) Les Biesecker, MD, National Human Genome Research Institute. Panelists reflected on the range of the term: "expertise" is used differently in the legal system, for the National Institutes of Health, within professional associations, and for medical doctors. An expert can be the parent of a child with an undiagnosed condition, a medical researcher, or a clinician.

Experts are needed to create and assess new, high-quality information, especially information based on new primary literature and case studies. Therefore, during the first few years of the ATCG project, there was much discussion around what constitutes expertise. If experts define "quality," how, as an industry, do we define "expert?" The key issues considered by ATCG were: "How do you determine who is an expert?" and, "Once 'expert' is defined, who will enforce the new criteria?"

When examined at a deeper level, the following questions emerged:

- Once we develop these criteria for what is high-quality information, who applies the criteria?
- How should these criteria be implemented?
- How do we update our tools?
- For whom/for what purpose are we producing these tools?
- How do we integrate financial matters?
- How do we address time and accessibility?
- What are the core endpoints?
- How do we define "credibility?"

From these topics, two were selected for further evaluation: How to define "expert" and "credible information," and the final question, "What are the core endpoints?"

The Consumer Perspective: Patients and Parents

Christine Vocke, whose son has an undiagnosed seizure disorder, became an expert on her child's condition and went on to become a lay professional with PXE International, an organization that provides information and support for people with pseudoxanthoma elasticum (PXE).

She described the diagnostic odyssey that began when her son had a seizure at age 4. While the simplest diagnosis was epilepsy, it became clear over time that his condition was more complicated. He developed new symptoms, and Vocke found that the doctors who treated her son preferred to focus on the seizures rather than the confusing new symptoms. There were experts on epileptic seizures, but none on her son's condition. So, she became the expert on viewing her son's symptoms in a holistic manner.

Vocke did not attend medical school, which often led to physicians dismissing her ideas. Some even refused to believe her account of her son's symptoms. Time was against her. As her child's health declined, she learned the vocabulary she should use to gain more credibility with physicians.

Vocke has the appropriate personal skills to persist and grow as a parent expert. For parents who are not comfortable in research and clinical settings, this is a greater challenge. One of PXE International's stated goals is to serve people who need help speaking with doctors and understanding research. This group disseminates "information for the PXE community," including patients, family, and doctors, to ensure there is credible and accessible information about PXE available to everyone needing it. An advocacy group like this is of particular importance to parent experts who may be treated with suspicion by the medical community.

The Researcher Perspective

Les Biesecker stated that for both experts and patients the question of understanding genetic conditions comes down to unraveling a tantalizingly simple question: what is causality, and what is mere association? One may see an event following another (i.e., a treatment and a change in condition, two physiological events) and believe that the first event caused the second. "We have deeply engrained ideas about cause and effect."

Understanding true science includes the ability to reflect on two events without assuming causation. This is critical for patients who are working against time to find answers about their condition and for researchers who are seeking answers in a poorly charted domain.

For Biesecker, then, an expert is someone who can correctly distinguish causality from correlation with respect to a condition, or determine that more information is necessary. Patients need to keep this distinction in mind when evaluating health information. Additionally, patients should be skeptical of anyone who claims to be "all knowing," as they are most likely frauds. On the contrary, individuals who collaborate with others, who build upon work that has been validated, are more likely to be a true expert in their field. "No scientist is an island."

The Provider Perspective

Howard Levy followed up on the question of who is a true expert, identifying different criteria that may serve well or poorly. He emphasized that someone who communicates well and delivers a favorable message is not necessarily an expert, even though there is a natural tendency to trust such a person. In the end, the best gauge of expertise is experience, and this "applies to clinicians, researchers, and lay people." However, with experience comes the risk of bias. People whose health improves after consultation with an expert will likely recommend the advice or treatment they received to others. On the other hand, those who did not have a positive experience are more likely to move on to another expert altogether rather than returning and giving the expert negative feedback. The best experts are open to novel approaches based on new, high-quality information.

Other criteria to consider when evaluating an expert are as follows:

- Education, training, and current or past affiliations can be helpful, although these criteria alone are insufficient to establish expertise or lack thereof.
- Expertise in one or a few areas does not automatically imply expertise in other areas.
- Credentials, titles, and positions of importance should be analyzed carefully as some may be perfunctory or volunteered.

Discussion

Moderated by Nancy Green

Different Kinds of Experts

- Generally, an expert is someone who has made a commitment to keep up with the literature in a field.
- Expert researchers can be defined by organizational affiliation with a recognized institution as well as their funding sources, the quality of their publications, the quantity of their speaking engagements on a topic, and by focused research on a disease.
- An expert healthcare provider can be characterized by experience (number of patients, years involved), professional interest (programmatic and institutional commitment), an interdisciplinary approach, recommendations by peers, a recognized institutional affiliation, and by speaking on a topic.
- An expert advocacy organization leader can be defined by the number of years involved in the organization, a willingness to partner, ability to stay up to date on information regarding the condition, effectiveness as facilitator of a community, ability to seek out evidence, and a broad and deep understanding of the research, clinical management and support issues, and the condition beyond the individual perspective to a population basis.

Panelists agreed that transparency is important to determine who should decide which experts should evaluate and characterize information for a condition. Audience members further asked, "Can family caregivers be considered experts on day-to-day living?" and, "Does having access to information make you an expert?" The attending family advocates and caregivers believed that yes, indeed, they are experts in lifestyle issues, and the present healthcare providers agreed. Everyone concurred that simply having information is not enough for any person to be considered an expert.

The Social Aspect of Expertise

Experts tend to organize themselves in a "club" that is closed to outsiders and to radical views. This may mean that the expert you need is inaccessible. Extremely well-qualified experts can reinforce this exclusion by referring people seeking information to their fellow experts who espouse a particular interpretation of the data, diminishing the chance that different views will be heard.

Grades of Expertise

The best question that can be asked about new information is "Who said it?" After that, the work of assessing a source's degree of expertise can begin. An audience member described levels of evidence and the way in which researchers rank the quality of data, ranging from an extremely scientific and verifiable quality (as in randomized clinical trials) to lay-person contributions, which are considered unreliable. In the case of rare genetic diseases, where there is so little scientific, high-quality data, more information is available at the anecdotal and lay-person level.

Conflicting Points of View

Experts will inevitably disagree. Biesecker stated, "Scientists have egos and that may lead to clashes." In a domain where there is so little irrefutable scientific data, scientists are forced to rely on other sources of validation: instinct, biases, hunches. When experts disagree, patients seeking information should not immediately take sides, and they should endeavor to avoid "bitter battles over things that don't make much difference."

Patient advocacy groups may also have different interpretations of the available data. One audience member proposed adding the concept of "spokesperson" to our understanding of what an expert is. A spokesperson is not the expert, but filters and transmits information from the expert to those who seek it. "People want an abbreviated, summarized version of information, and so the spokesperson is crucial." Sometimes just being told what is not yet known is the most valuable information a patient receives.

Medical students can also be a good source of new information, but some people are wary of the information they bring, because students may simply regurgitate what they learned, rather than bring their original thinking. The audience suggested that courses in critical thinking become emphasized as part of the educational process so that the next generation will be better prepared to assess rapidly changing information.

Expertise as Team Work

In the end, the discussion evolved to a different way of defining expertise. The expert clinician can be wrong, especially in an area like genetics, where there is so much missing information. The patient can be wrong, as he does not have the medical training to accurately theorize about his condition. Medicine should therefore be "a team activity," obviating the need to define and identify experts. The best solution may not be one person with all the answers, but rather several people working together, respecting each other's views, and developing a relationship where problems can be worked out productively. This means that doctors need to listen to and respect patients, and patients need to trust the doctors they choose.

Information as a Moving Target

There is much to be learned about genetics, genetic conditions, and health. It is hoped that more information about these conditions will be discovered as advances are made in genetic understanding, or "Genome 2.0." These rapid advancements necessitate that the researchers and the advocacy community be similarly quick to ask and address the appropriate questions about ethics and safety.

PANEL DISCUSSION: CONTENT

Kate Reed MPH, ScM, CGC, National Coalition for Health Professional Education in Genetics (NCHPEG), introduced the topic, moderator Joseph D. McInerney, MA, MS, NCHPEG, and the panelists: 1) Amanda Bergner, MS, CGC, Johns Hopkins University; 2) Jayne Dixon Weber, National Fragile X Foundation; and 3) Ben Heywood, MBA, Patients Like Me. The panel discussed the following questions: How should we choose appropriate information for the audience? What is the best way to organize the information for maximum effectiveness? Given the volume of emerging information and the multiple ways to access it, these two issues of content and organization are inexorably linked.

Theory: How Do People Think About Information?

Amanda Bergner discussed how patients perceive medical information related to their diagnosis. She introduced grieving as part of the process of understanding a diagnosis.

It is important to remember that each individual patient may experience different levels of fear or anxiety, and that receiving a diagnosis can be very painful. To better understand a patient's reaction to a diagnosis, genetic counselors use theories of grief and mourning. Understanding a scientific concept and actually hearing a diagnosis for the first time are entirely different experiences. The latter experience often involves "giving up an understanding of who you thought you were or who you didn't think you were." Grieving is natural and common.

Bergner explained that people grieve in different ways that can impact their understanding and perception of information. While grieving styles can be imagined to exist on a continuum, Bergner asserts that two types of response fall at the ends of the continuum:

- *Instrumental* grievers are "doers" who process grief by actively responding against the diagnosis, with "a voracious appetite for information." Providers may assume that these people are coping well because of their orientation toward information, but this is not always the case. Accumulating information can be a means by which they avoid the emotions arising around receiving a diagnosis.
- *Intuitive* grievers are information-averse, and they direct their energy into emotions. They are not often well-equipped to process detailed information, and often respond to new information by crying or wanting to talk. They need to connect with others to process what the diagnosis means to them.

There are generally two phases to any patient's processing of this medical information, no matter where she is on the grieving spectrum:

- The "deconstruction" phase is a vulnerable time when people give up their prior sense of self. Typically during this period patients do not want any new information because they are undergoing a critical psychological transition.
- The "reconstruction" phase signals the beginning of patient's receptivity to new information. The patient becomes "more rational" and seeks information that will help him understand the diagnosis better.

Healthcare providers who focus on the content of the diagnosis rather than recognizing the emotional impact on the patient will miss the opportunity to establish effective communication at a critical point in the patient's life and in the provider-patient relationship. Therefore, when diagnosing a genetic disorder, whether the provider is expert or poorly versed in its details, it is important for that provider to focus more on the patient rather than the diagnosis. This will provide a foundation for future communication with the patient as he moves forward into the period of acceptance.

Practice: What Advocacy Groups Need to Communicate

Jayne Dixon Weber has a son who was diagnosed with Fragile X syndrome 20 years ago. As a parent and lay expert she discussed how to best present information to patients and caregivers through a disease advocacy organization website, using the National Fragile X Foundation's website as an example.

Dixon Weber was pregnant with her second child when she received a diagnosis for her first child's condition. Like Vocke of PXE International, she had the necessary personal skills to navigate the diagnostic odyssey efficiently. Being an engineer, Dixon Weber was comfortable processing complex scientific information and communicating with experts in a way that increased the chance they would treat her insights with respect.

She has found that many parents ask the same questions and suggested that the medical community is not adequately answering these questions. Often parents turn to the Internet for answers, where information quality is variable. Dixon Weber's comments on the quality of information available online echoed earlier comments: the critical question is not what is presented but, "who said it?"

Dixon Weber described goals and concerns in developing educational materials for Fragile X syndrome similar to those described with regard to the Duchenne muscular dystrophy website (see Panel Discussion on Content).

- An effective website about a genetic condition must communicate new medical information and serve the emotional needs of the community. Patient stories can fulfill both these goals.
- To ensure information quality, advocacy groups should review user-submitted information before it is published on the site.
- Educational materials should take into account the way in which the audience is processing the information.

In developing new content and thinking about future developments, Dixon Weber endeavors to improve the user experience. She imagines creating a feedback system to measure how people are using the website and in what capacity.

Vision: The Next Level Shift in Information about Genetics

Ben Heywood described the risks and rewards of sharing both personal information and emerging medical data on a for-profit website. Heywood's brother had amyotrophic lateral sclerosis (ALS), which was the impetus for starting Patients Like Me. Information about his brother's experience is included on the website for other patients with ALS. Consumers come to Patients Like Me with common questions: "Given my status, what is the best outcome I can expect to achieve, and how do I get there?" Patients Like Me is dedicated to providing patients with the best possible information about their condition, using the experiences of other patients with the same condition.

Patients Like Me relies on several key principles:

- Openness: Patients openly share health information, understanding that the benefit of learning about others' experiences is balanced by the risk of potentially acting on this information without their doctors' approval.
- Transparency: Patients Like Me is explicit about what they do with information collected on the website and their opinion of the safety of providing such information in this format.
- Community focus: Similar to previously described advocacy websites, Patients Like Me is not only about data, but about emotional support.
- "Auditable" sources: The website allows consumers to trace each specific point of information to its source, answering the "who said it?" question.
- Full access to all data: Patients Like Me shows historical data as well as current data to allow consumers to draw their own conclusions.
- Deliberate conclusions: This site slows down the dissemination of the most current data so that people can review it carefully before posting it.

Heywood imagines a new and ambitious use for Patients Like Me—to accelerate discovery by collecting data particular to specific disorders on the site. This patient-driven collection could allow for a longitudinal study of populations with specific conditions who are not being treated at the same hospital or even in the same country. Their only common factor would be their shared disorder. Treatments and outcomes could be tracked and compiled for research.

Discussion

Moderated by Joseph D. McInerney

Who Is Reading Your Webpage?

- Reading Ability: Some websites about genetic conditions require a level of literacy that is much higher than is needed to use Facebook, for example. Successful websites consider user literacy. Not all parents of children with a genetic condition speak English as a first language or have a degree in science. Understanding your target audience can help focus your efforts.
- Computers: Some patients or caregivers may not have access to the Internet. At the National Fragile X Foundation, newly diagnosed patients can receive printed "special topic brochures" about specific issues. Additionally, new members can call for information.

Health Status

Patients Like Me users share anecdotal evidence. Because of the self-selected nature of the contributors, this evidence may not give a global picture of the condition. Heywood notes, "It is possible that individuals who feel better don't participate… We clearly think that we are going to get people who are not responsive to drugs, and are dealing with other issues every day." Thus, the information on the site may be biased. Similarly, any system that filters and prioritizes information will have an inherent bias.

The Moment of Truth: Delivering Bad News

- Audience members emphasized the importance of training medical students to sit with healthcare providers delivering diagnoses to learn this critical skill.
- Bergner reiterated the psychological impact of receiving a diagnosis: "Some families are not ready for all the information and some are." Dixon Weber elaborated, based on her experience talking to parents of children with Fragile X syndrome: "Talking to families with a 3-year-old child is very different from talking to families with a 2-week-old baby." Delivering a diagnosis to a patient should include a conversation with the whole family.

Knowing What You Do Not Know

In contrast to the admonition that some patients may not be ready for a lot of information about their disorder at the time of diagnosis, some speakers asserted that healthcare providers have a responsibility to share large amounts of information. They should tell patients not only the basic facts about their disorder, but also anticipate the questions that patients should ask but are unable to imagine. "Road map" was a commonly used metaphor to describe the evolution of a patient's understanding of his genetic disorder. The doctor or lay advocate knows the map based on their experiences with the progression of the disorder; the newly diagnosed patient, though, is unfamiliar with the map and needs to be coached about the types of questions he will need to ask and answer along the way.

The audience discussion can best be summed up in the words of one speaker who said that what families need is "information with information on how to get more."

PANEL DISCUSSION: USABILITY

Meredith Weaver, PhD, ScM, CGC, University of Maryland School of Medicine, introduced the panel on usability of genetics information, moderated by Alejandra Gepp, MA, National Council of La Raza: 1) Paula Raimondo, MLS, Health Sciences and Human Services Library, University of Maryland; 2) Therese Ingram Nissen, MA, NCHPEG; 3) Mark Boguski, MD, PhD, Harvard Medical School/Beth Israel Deaconess. Weaver echoed that patients often need to emotionally process a diagnosis before they can absorb new information and act accordingly.

How People Interact with Information

While all patients need to process information about their diagnosis emotionally, many patients may be at a disadvantage if they have low health literacy, according to Paula Raimondo. She cautioned about the danger of a healthcare provider overestimating a newly diagnosed patient's ability to understand medical information. It is the provider's responsibility not only to appreciate the psychological process by which a patient apprehends his or her diagnosis, but also to assess the patient's health literacy.

Even average literacy is generally insufficient to understand medical materials. For example, while the average adult reads at an 8th-grade level, most health-related materials are written at a high school level or higher, meaning they are too difficult for the average adult to comprehend. Health-related materials written at the high school level are entirely out of the reach of the 20% of the population who read at a 5th-grade level or below.

The lack of health literacy in the population has medical consequences. Illiterate or barely literate adults have coping mechanisms for navigating in a literate world. It may not be obvious that a patient cannot understand a brochure written at a high school level. Illiterate patients may feel stigmatized and be unwilling to confide that they cannot read what the doctor or receptionist hands them. Such patients, who are mostly older, immigrant, and non-white, are "less likely to seek help early, complete forms correctly, follow medical regimens, keep appointments," Raimondo said. As a result, their disorder may progress more rapidly and further than it would in a highly literate patient who is able to engage in chronic disease management at an early stage.

Taking into account a patient's health literacy is one more layer of complexity in the process of communicating a healthcare "road map" to the newly diagnosed patient.

Developing Web Pages

To design a user-friendly website for patients, Therese Ingram Nissen recommended seeking consumer feedback at the beginning of the design process. "Involve your audience early and often" was the main theme of her guidelines for effective Web design, a message that is particularly apt for patient advocacy websites.

Good Web design does not start with the designer but with the users. Nissen advised that developers first determine the basic criteria for the site, relying partly on information gathered by interviewing potential users, caregivers, and health experts. Then go back to them with a preliminary design and ask for feedback. The goal is to create a "scenario of use." The best websites will create profiles of users so that the site can be easily customized by user type; patients have different needs than physicians. Caregivers have unique needs when they are searching online for information and support. Nissen echoed what other speakers said about the Web: it has been user-driven, and now it is increasingly user-created. High-quality Web design involves an active investigation into how consumers use the information they find in their everyday life.

She outlined some specific details to bear in mind when creating a new website.

- Users read online material 25% slower than they read a book, so present only about half the content you might present in a brochure.
- Users read online content in an "F" pattern: they look at the heading (the top line of the F), then glance at the subheading (the smaller line), and may go on to read the first two sentences. Use this structure to

organize information so that users can navigate easily, quickly finding what they need or moving on to a page that is more relevant. Break up the text with bullets, and use graphics, tables, and even white space to provide a comfortable and efficient display.

Web design today, according to Nissen, is not so much about technology as it is about how people process information. This is Web 3.0.

Social Media and Disseminating Information about Genetics

While other speakers described the ways in which human knowledge and technology are increasingly dovetailing, Mark Boguski pointed out several incongruities that persist, which contribute to the "digital divide" we live in today.

According to Pew Internet surveys, 160 million people use the Internet for health information, but many do not disclose this fact to their healthcare providers. The Internet clearly satisfies a patient need, but leads to a doctor-patient relationship that is not based on clear and open communication.

Those who use the Internet for health information may not be using it as effectively as possible. Something as simple as taking notes may make a person less efficient at Web-browsing. "Most people using Google still use pencil and paper to notate things about websites" instead of switching back and forth between the website and the computer's notepad to make notes directly in a document.

Boguski described a new online medical search environment, resoundinghealth.com, designed to enhance the experience of seeking health information on the Web. "Casebook," for example, allows users to put aside pencil and paper and type notes online in a pdf. Resoundinghealth.com aims to "store, organize, and share personalized results of online searches" as efficiently as possible.

Boguski also detailed steps for using social media to disseminate medical information. A "patient with a textbook symptom is an opportunity to take textbook information and put a face on it." But this face cannot be static—it needs to emerge through "dynamic and socially-constructed interactions." How is this done?

- Social media must first gain users' attention. One example of a site with a good "hook" is celebritydiagnosis.com. The blog features celebrities' health issues and provides links to information about the diseases that are mentioned.
- To keep users' attention, the site's material should be engaging.
- The site should provide "learning guidance," a similar concept to the "road map" described earlier.
- Users should have the ability to rate different parts of the site and provide feedback.
- The site's presentation should enhance information retention and encourage users to go deeper into the material.

Websites that provide information to healthcare consumers have a responsibility to educate, not just inform, those consumers

Discussion

Moderated by Alejandra Gepp

Processing Information

Presenting information to a newly diagnosed patient is a complicated process, involving players whose needs and wants are often at odds. "It's about how much your client needs/wants to know." On one hand is a doctor, with her set of experiences and education, building a road map based on the needs and symptoms of the patient, offering answers to questions that the patient needs to know. On the other hand is the patient, processing a diagnosis emotionally, with a set of information that he wants to know (or explicitly does not want to know). Bergner fully elaborated this concept in her comments about the two types of grieving that different patients experience.

The Digital Divide

There are many types of literacy at play in understanding health information. General literacy is an important factor, but one audience member noted the difference between general literacy and health literacy, and digital literacy is yet another factor. Speakers described health-illiterate patients with low general literacy or a lack of computer skills. The participant countered that his father is highly literate, but nonetheless has trouble understanding genetics articles.

Similarly, participants commented that hyperlinks are useful for people who are highly computer literate, but useless for people who read on the Web as if they were reading a brochure. For these users, printable versions should be available in a way that preserves the hyperlinked information.

The need to communicate with all members of a community affected by a genetic disorder requires using a variety of tactics. We cannot "leave behind a subset of the population." In general, Gepp concluded, when "designing educational materials or in communicating information, I suggest using a combination of print and Web-based formats."

NEXT STEPS

Terry posed the question, "How do we give people the tools to decide if something they are reading should be trusted or trashed?" She proposed a button that consumers would click on to evaluate information. The button would allow consumers to see the answers to three critical questions:

- Who said it?
- When did they say it?
- How do they know?

The answers to the questions would help consumers decide whether to trust or trash the material. Audience members returned to the idea that both information and the delivery format are moving targets.

Information as a Moving Target

"How do you give consumers tools when the content isn't static?" asked Heywood. One speaker suggested refining "trust it or trash it" to "trust it, hold it, or trash it." This would allow users to rate "a good study in science that hasn't been replicated yet," for example. A preliminary assessment might rate the study as "good" rather than something to be trashed, but it would not receive a "trust it" designation until further work confirms the findings.

Delivering Information

Participants agreed that the delivery format of information should evolve and adapt with the changing needs of its users, which again brought up questions of computer use. If the Web is the best, most customizable format to deliver information, how do you reach patients who are not computer literate? The degree of computer literacy in the population is expected to change over time. Children in elementary schools in America are being taught how to "access, use, and corroborate information" online while their parents may never have used computers in school. Terry compared going online today to having a cell phone when they were first introduced. Many people who said then "I will never have a phone in my pocket" are today avid smartphone users. The interface is evolving and so is the user population. But for now, how can today's consumers best be accommodated?

Who Said It?

Sometimes the answer to this question is not "who" but "what," as organizations, rather than individuals, are often sources of information. On the other hand, a personal voice can carry weight in a way that the opinion of an organization cannot, such as with thoughtful peer reviews. The question "who said it?" should therefore involve taking into account the different advantages of an organization's view and a personal testimonial.

Who Reads It?

The audience had conflicting opinions as to how to present information at the right speed. There must be a balance between getting information out quickly enough to reflect current findings for patients whose needs are time-sensitive, but slowly and thoughtfully enough to allow researchers to evaluate new research and present information with greater confidence of its accuracy. Participants concluded that the Web is not quite capable of delivering high-quality answers to patients in "urgent situations."

Some medical website users seek to assert their own agendas rather than search for the best information. This is a significant challenge, and the audience was stumped on how to address it. Sensitive user-submitted information, such as patient blogs, are particularly vulnerable. However sophisticated Web 3.0 may be, protecting patient privacy while disseminating information remains a challenge.

DINNER DEBATE: THE WISDOM OF CROWDS

Terry introduced the concept of "the wisdom of crowds" popularized by James Surowiecki in the bestselling book of the same name. A crowd of unrelated people may have a type of collective wisdom, whether the crowd is a dangerous mob or a group of disparate scientists eventually coming to a commonplace solution. The wisdom of crowds does not come from a single source: "a well-placed propaganda campaign is not the wisdom of crowds." It is fluid and evolving.

With the emergence of Web 3.0, whether we like it or not, the crowd is already disseminating and validating information about genetic conditions. Some members of this crowd are patients and caregivers, some are medical experts, and some have no first-hand knowledge about genetic diseases. What role does the wisdom of crowds play in accessing credible health information?

Boguski, Heywood, and Barris made statements about tapping into the wisdom of crowds in the context of user-friendly, patient information delivered electronically. Boguski and Barris illustrated opposing views of the wisdom of crowds. On the one hand, Boguski argued that "with the large amount of information being produced, at the end of the day, the information that is not valid or even fraudulent won't be used"; the wisdom of the crowd will serve to invalidate it. "The truth will prevail." On the other hand, Barris pointed out that the time and amount of input that a crowd needs in order to invalidate bad data could cost a patient his life. "At any moment in time, theoretically, someone could write something on a biomedical page that killed someone—even if 16 seconds later the information could have been reversed." The concept of a 16-second dissemination of fatal data reverberated throughout the discussion.

Playing devil's advocate, Heywood proposed that some good could come from the 16-second dissemination of fatal data. If there is a brief and quickly quashed flash of data that could result in a patient death, "who cares?" He suggested that the one-time transmission of erroneous data could accidentally yield information that might in some way advance understanding of treatment for a disease like ALS. Currently, the life expectancy for ALS patients is two to five years after diagnosis. But what if that 16 seconds ultimately speeds up the cure for ALS by one year? That cure will affect the approximately 30,000 Americans living with ALS today.

Mitigating the 16-second Risk

- One idea to eliminate the 16-second risk would be for a governing body like the National Institutes of Health (NIH) to give grants to medical experts to oversee websites that contain medical information. With information proliferating rapidly, clinical geneticists cannot spend more time policing public websites. This grant system would hopefully allow experts to catch inaccuracies quickly, and would cut down on false information being disseminated. However, this does not ensure that a medical expert will always make the right correction or have the unique knowledge needed in each case.

- Absent an NIH grant, some audience members believed it to be the responsibility of the patient advocacy community, with vast knowledge and understanding, to keep a constant eye on the accuracy of medical information being disseminated.
- Participants largely agreed that medical students should not use consumer websites for authoritative information. One audience member asserted that "Clinicians need to take hold of our own game and say 'no,' we are going to train our residents to look at peer-reviewed literature, and not Wikipedia or such sources."

Audience members discussed the image of an insiders' circle of experts delivering the information they deem pertinent to an outside circle of patients. However, Web 3.0 may change the traditional role of the patient. Barris summed it up: "The public has a responsibility to be the ones on the front lines: if you think you have a solution, then be bold." Get your idea out to the experts. Another person countered, "No wisdom of crowds will ever identify a gene for a condition."

The roles of scientific data consumers and scientific data producers are evolving in jumps and starts; these changing roles suggest rethinking how information is delivered online. Heywood proposed turning the way we thought about disseminating information upside down: Rather than evaluate information and then disseminate it, we should work backwards to "figure out how the information is being consumed, where it is being consumed" and then determine what people should know. Discover what they are looking for and deliver that. This concept represents a radical departure from the image of an insiders' circle delivering information to outsiders. In this vision, the online behavior of the outside circle of patients would dictate what information would be delivered to them from scientists.

The Web 3.0 medium complicated the picture: information that can be delivered is determined by the format of the websites consumers are actually using. As Greene put it, "you have to go where people are reading." This idea brought the discussion back to the question, "Who is reading it?" What is the profile of consumers using online information at a given website?

There are two extremes of information delivery via the Web. At one extreme, information for patients is delivered in some type of free-for-all with the assumption that over time, "the truth will prevail." At the other is a Web in which information is censored and delivered selectively, scientific information to scientists and subjective information to consumers. Ultimately there may be a safe and morally responsible middle ground. Instead of "filtering" the wisdom of crowds to parse out what may be dangerous, we may need to "focus" the wisdom of crowds to achieve the best possible use of that wisdom. Terry concluded the discussion by sketching the idea of a "gate-kept" wisdom of crowds for credible information about genetic conditions.

CONCLUSION

This energetic meeting was an excellent forum for discourse with diverse stakeholders about the need for credible information. The availability of information is increasing at an unprecedented rate. In order for healthcare consumers to effectively manage their own health, they must be empowered to discern the quality and relevance of available information. Especially with today's social media and the inevitable developments of Web 3.0, we must all vigilantly examine the credibility of information on an ongoing basis.

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ACRONYMS

ACCE: Analytic validity, Clinical validity, Clinical utility, Ethical, legal, and social aspects

ATCG: Access to Credible Genetics Resource Network (www.geneticalliance.org/atcg)

DAO: Disease Advocacy Organization

EGAPP: Evaluation of Genomic Applications in Practice and Prevention

GWAS: Genome-Wide Association Studies

NIH: National Institutes of Health

NBS: Newborn Screening

SNP: Single Nucleotide Polymorphism

GLOSSARY

Analytic validity: How accurately and reliably a laboratory test measures a particular genetic characteristic.

Blog: Short for weblog, a type of website usually maintained by an individual with regular entries of commentary, descriptions of events, or other material such as graphics or video.

Clinical utility: How likely an intervention or test is to significantly improve patient outcomes.

Clinical validity: How consistently and accurately a test detects or predicts the clinical status of interest.

Diagnostic odyssey: An extended and laborious effort to obtain a diagnosis for an unknown disease or condition.

FRAXA Research Foundation: (www.fraxa.org) An organization whose mission is to accelerate progress toward effective treatments and a cure for Fragile X syndrome by funding the most promising research.

Genetic counselor: Health professionals with graduate degrees and experience in medical genetics and counseling. Genetic counselors provide individuals and families with information about genetic conditions, help them make informed decisions, and provide psychosocial support.

Genome 2.0: A new picture of how the genome works as a whole, with a shift in focus from single protein-coding genes to understanding interactions within the entire genome.

Human Genome Project: A 13-year effort coordinated by the U.S. Department of Energy and the National Institutes of Health to identify and sequence all of the genes in human DNA.

Hyperlink: A word, phrase, or image on a website that the user can click on to jump to a new document or webpage, or a new section within the current page.

National Coalition for Health Professional Education in Genetics (NCHPEG): (www.nchpeg.org) A coalition of organizations that works to improve the healthcare of the nation through promoting health professional education and access to information about advances in human genetics.

National Council of La Raza: (www.nclr.org) A civil rights and advocacy organization that works to improve opportunities for Hispanic-Americans.

National Fragile X Foundation: (www.fragilex.org) An organization that unites the Fragile X community to enrich lives through educational and emotional support, promotes public and professional awareness, and advances research toward improved treatments and a cure for Fragile X.

National Institutes of Health: A part of the U.S. Department of Health and Human Services, the primary federal agency for conducting and supporting medical research.

Parent Project Muscular Dystrophy: (www.parentprojectmd.org) An organization that improves the treatment, quality of life and long-term outlook for all individuals affected by Duchenne Muscular Dystrophy through research, education, advocacy and compassion.

Social media: Websites that allow users to have a conversation online by sharing content and media.

Web 2.0: A term that refers to Web applications that facilitate interactive information sharing, interoperability, and collaboration, as opposed to Web 1.0 which references read-only content and static websites.

Web 3.0: A term that refers to the semantic Web (an evolving development of the Web in which the meaning of data is defined, making it possible for the Web to "understand" and satisfy the requests of people and machines to use the Web content), personalization (e.g. iGoogle), intelligent search, and behavioral advertising.

Wiki: A collaborative website that allows anyone to edit, delete or modify content that has been placed on the site, including the work of previous authors.

Trust It or Trash It?
A tool to help evaluate and create genetics health information

What is the *Trust It or Trash It?* tool?

Trust It or Trash It? is a free online tool that serves two purposes: to encourage critical thinking as people encounter health information and to add to the existing volume of high quality genetics materials. There are two versions of the tool, one for people who are developing educational materials and one for people assessing the quality and completeness of health information. The version for judging the quality of health information (www.trustortrash.org) starts with three basic questions: Who said it? When did they say it? How did they know? These questions help the reader think about the information critically. The second version, for developers, provides additional guidance for choosing appropriate content and effectively presenting information (www.trustortrash.org/developer).

Who made the *Trust It or Trash It?* tool?

The tool was created as part of the Access to Credible Genetics Resources Network. Condition-specific advocates, healthcare providers, genetics professionals, and health educators contributed to the tool's development and pilot testing.

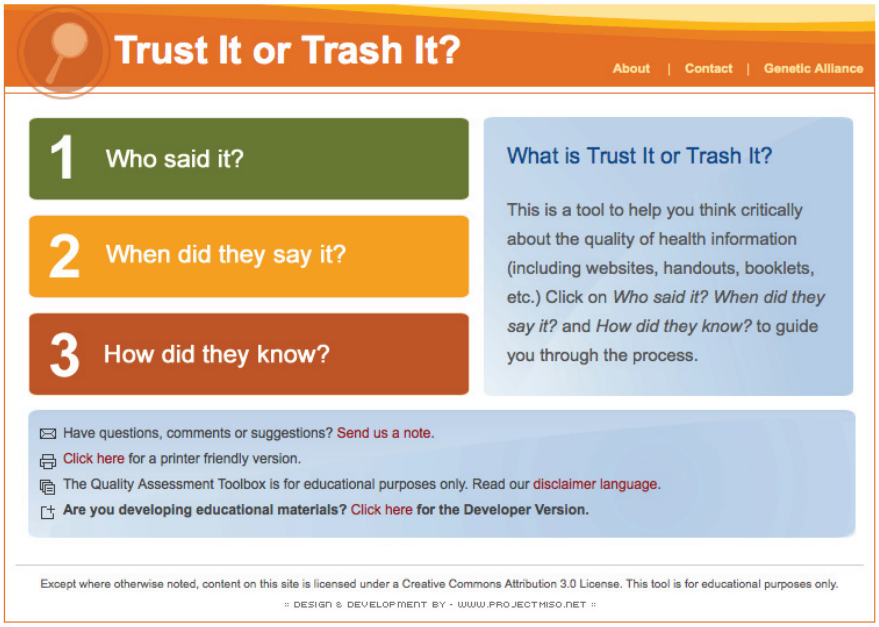
Why is the *Trust It or Trash It?* tool needed?

Health information is available from a wide variety of sources, some of them trustworthy, some of them not. Determining which pieces of information to believe can be very difficult, especially when the information is new to you. While there are guidelines to determine the quality of some medical treatments, there is no similar guidance to help people judge whether information is good or bad. This tool helps fill that gap.

What's next?

- A "widget" that can be hosted on any website will be available. The "widget" is an application that will open the tool as a static horizontal bar that stays open on the bottom of the computer screen. This application will allow users to evaluate the content on their computer screen without having to visit an outside website.
- Both versions of the tool will be translated into Spanish.

Trust It or Trash It?
A tool to help evaluate and create genetics health information



Trust It or Trash It?

1 Who said it?

2 When did they say it?

3 How did they know?

What is Trust It or Trash It?

This is a tool to help you think critically about the quality of health information (including websites, handouts, booklets, etc.) Click on *Who said it? When did they say it?* and *How did they know?* to guide you through the process.

Have questions, comments or suggestions? Send us a note.

Click here for a printer friendly version.

The Quality Assessment Toolbox is for educational purposes only. Read our disclaimer language.

Are you developing educational materials? Click here **for the Developer Version.**

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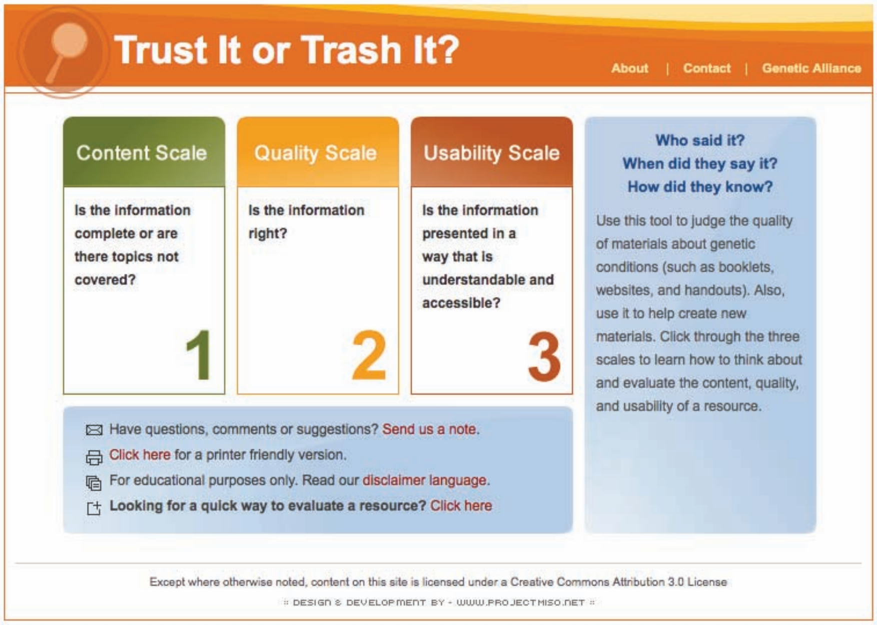
‹‹ trustortrash.org

This version encourages critical thinking as people encounter health information.

Project Partners:

- National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention
- Genetic Alliance (lead)
- FRAXA Research Foundation
- GeneTests
- National Coalition for Health Professional Education in Genetics
- National Council of La Raza
- National Fragile X Foundation
- Parent Project Muscular Dystrophy
- University of Maryland School of Medicine



Trust It or Trash It?

Content Scale	Quality Scale	Usability Scale
Is the information complete or are there topics not covered? 1	**Is the information right?** 2	**Is the information presented in a way that is understandable and accessible?** 3

Who said it?
When did they say it?
How did they know?

Use this tool to judge the quality of materials about genetic conditions (such as booklets, websites, and handouts). Also, use it to help create new materials. Click through the three scales to learn how to think about and evaluate the content, quality, and usability of a resource.

Have questions, comments or suggestions? Send us a note.

Click here for a printer friendly version.

For educational purposes only. Read our disclaimer language.

Looking for a quick way to evaluate a resource? Click here

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‹‹ trustortrash.org/developer

This version assists the development of quality educational materials.

How do I get more information?

Contact Amelia Chappelle, Associate Director of Genetics Resources and Services, Genetic Alliance, 202.966.5557, achappelle@geneticalliance.org, **www.geneticalliance.org/atcg**

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